

Course description

This course will introduce basic concepts of human evolution and population genetics – the processes which created the etiological architecture of complex human disease in today's population. We will explain why most human phenotypes are under the influence of an enormous number of (mostly individually rare) genetic factors which vary both within and between populations. The failures of genome-wide association studies to identify important risk factors for common disease will be shown to be completely consistent with what evolutionary theory actually predicts and is likewise consistent with real data from genetic studies in agriculture and model organisms. Because a researcher cannot do genetic experiments in humans but is forced to work with observational data, we will further discuss how the researcher searches for and utilizes natural experiments that approximate the studies we would design if we had a more convenient species to study. Since evolutionary theory is so critical to understanding exposure distributions in genetic variation, it is critical for anyone working in genetic epidemiology to have a conceptual background in genetics.

Lecturers

- ▶ Joseph D. Terwilliger, PhD (Columbia)
- ▶ Joseph Lee, DrPh, MPH (Columbia)
- ▶ Kenneth M. Weiss, PhD (PennState)

Information and registration

www.tmf-ev.de/school-extra

Infrastructures for Medical Research

The TMF is the umbrella organization for networked medical research in Germany. It is the platform for interdisciplinary exchange as well as cross-project and cross-location cooperation in order to identify and solve the organizational, legal/ethical and technological problems of modern medical research. Solutions range from expert opinions, generic concepts, and IT applications to checklists, practical guides, training, and consultation services. The TMF makes these solutions available to the public free of charge.

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Logical Reasoning in Human Genetics

September 24–28, 2012
Berlin | Germany

TMF – Technology, Methods, and Infrastructure
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Logical Reasoning in Human Genetics



Agenda

Philosophical and Evolutionary Background

- ▶ Introduction: What is ›logical‹ about this course or about science?
- ▶ Doubt: Scientific inference in a probabilistic world
- ▶ Evolution and the Darwinian method: Why should traits be ›genetic‹?
- ▶ »The human genome« doesn't exist. Why is it so useful? Genome evolution

Simple traits?

- ▶ The consequences of being diploid: »Mendelian« inheritance
- ▶ Just like peas! Genetic epidemiology of simple traits
- ▶ But ... not simple like peas after all
- ▶ Hunting causes of simple traits: linkage and LD analysis – theory and applications

Complex traits and the evolution of genetic architecture

- ▶ Phenogenetics and the consequences of evolution by phenotype: polygenic traits
- ▶ Environmental and cultural cofactors and the like
- ▶ Detectance, study design, and linkage and LD analysis of complex traits
- ▶ The genomic ›commons‹: a cornucopia of public tools and resources
- ▶ Simulation is poor man's theory by another name: ForSIM

Applied genetics (study design and data)

- ▶ Genetic Epidemiology approaches to complex traits
- ▶ Study designs for complex traits
- ▶ Applications of linkage and LD analysis to complex traits
- ▶ Designing your study: simQTL as a simulation aid
- ▶ Effect size estimation, attributable risk, and their equivalents in genetics.
- ▶ ›Hidden heritability‹ isn't hidden: the complex nature of complex traits
- ▶ Not all inheritance is Mendelian, but it's all evolutionary (somatic mutation)

GWAS, Biobanks, and Biotechnology to the rescue?

- ▶ Historical overview of GWAS and HapMap: GWAS, Biobanks and other convenience studies for complex traits
- ▶ GWAS and Biobanks: we see what we should expect to see (unfortunately)
- ▶ Natural Experiments in human genetics – a better alternative to GWAS
- ▶ Revisiting truth: Statistical inference of statistical causation
- ▶ Wrap-up, Summary, and Discussion

Organisational issues

Date

September 24–28, 2012

Venue

TMF Conference rooms | Georgenstraße 22 | 10117 Berlin, Germany

How to get there

- ▶ **By rail:** from Berlin main station (Hauptbahnhof), take the S-Bahn (suburban rail service) and alight at Friedrichstraße.
- ▶ **By air:** from Tegel airport, take the no. 128 bus to Kurt-Schumacher-Platz. Then take the U6 (underground) to Friedrichstraße. From Schönefeld airport take the Railway to S-Bahn station Friedrichstraße.

The venue is only a short walk from the Friedrichstraße station (map see: <http://www.tmf-ev.de/EnglishSite/AboutUs/Contact.aspx>).

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Registration

Participants: 25–30
Registration fee: € 700,00
Online registration:
www.tmf-ev.de/school-extra